

FIGURE 1

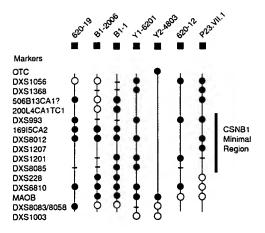
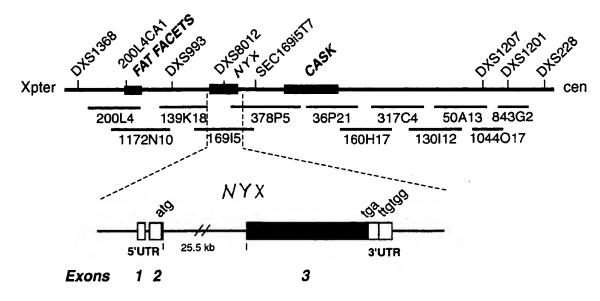


FIGURE 2

a Physical map of the CSNB1 minimal region and genomic organization of NYX



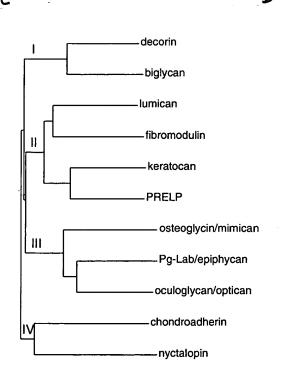
b Leucine-rich repeats and the distribution of mutations in the Nyctalopin protein

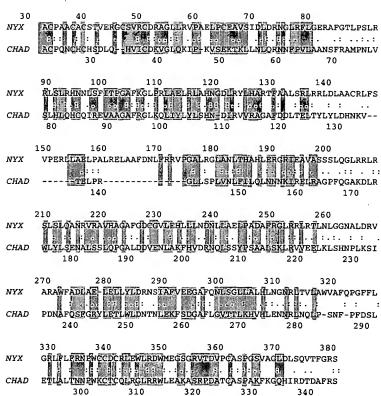
MKGR	emlvlllhavvlglpsawavga a <u>ra :paa ::a</u> :stverg :svr :dr	-	50
	del		
1.	AGLLRVPAELPCEAVSIDLDRNGL	-	74
2.	RFLGERAFGTLPSLRRLSLRHNNL	-	98
3.	SFITPGAFKGLPRLAELRLAHNGDL	_	123
4.	RYLHARTFAALSRLRRLDLAACRL	-	147
5.	fsvperllaelpalrelaafdnlfrrvpgalrgl	-	181
6.	an <i>l</i> thahl15lrrlrsls <i>l</i> qanrv	-	218
	ERGRIEAVASSSLOG		
7.	ra v haga f gdcgv <i>l</i> eh l l l nd n l l	_	242
8.	AELPADAFRGLRRLRTLNLGG <i>N</i> AL	_	266
9.	DR v arawfad l aelelly <i>l</i> dr n si	-	290
10.	af v eega <i>f</i> onlsgllalhlngnrl	_	314
11.	TV L AWVA F QPGFF L GR L F L FR N PW	_	338
	β -sheet α -helix		
	· 1		
JEDE	RLEWLRDWMEGSGRVTDVP TASPGSVAGLDLSOVTFGRSSDGLCVD	_	388
PEELI	NLTTSSPGPSPEPAATTVSRFSSLLSKLLAPRVPVEEAANTTGGLA	_	438
NASL	SDSLSSRGV GGA GROPWFLLASCLLPSVAOHVVFGLOMD	_	481

A insertions of SVPERLL, GLR and RLR, respectively

v most likely signal peptide cleavage site

FIGURE 3





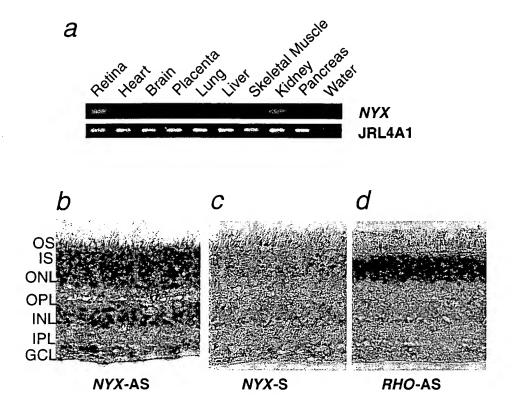


FIGURE 4

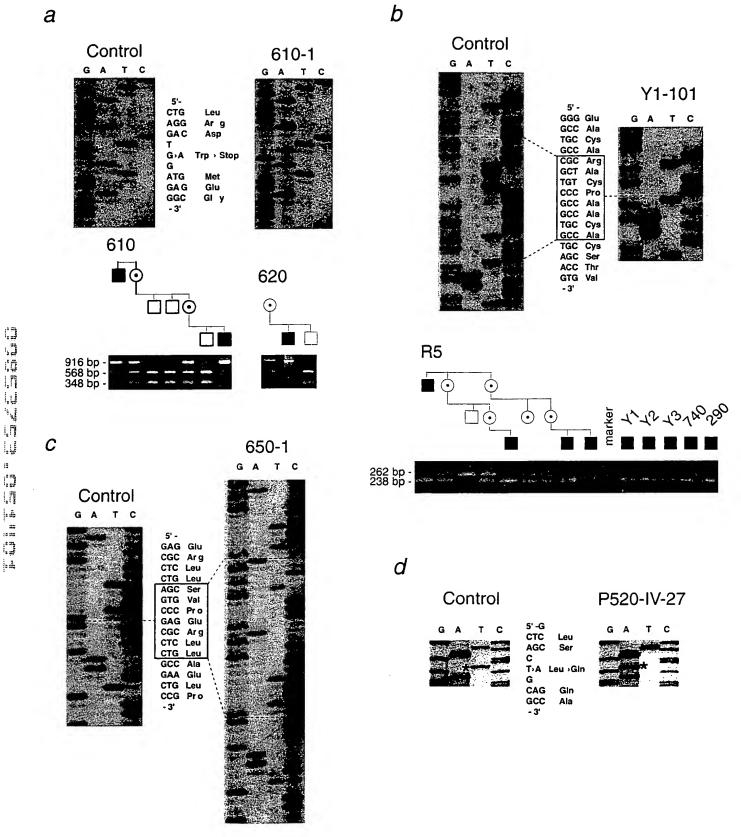


FIGURE 5

Table 1 • Nyctalopin mutations in families with complete CSNB

Family ^a	Origin	Mutation ^b	Codon change	Predicted effect on nyctalopin
290.740.830.Y1.Y2.Y3.R5°	USA	85-108del24nt ^d	RACPAACA29-36del	partial loss of N-terminal cysteine cluster
P23.340 (2) (ref 13) Neth	Netherlands 452C>1	452C>T	P151L	missense, proline to leucine
650 (2)	Canada	464^465ins21nt	SVPERLL155-156ins	expansion of LRR5 ^e
750 ^f (1),780 (1)	Canada, USA 551T>C	, 551T>C	L184P	missense, leucine to proline
540 (6)(ref 28, family 2)	Germany	556-618del50ins3nt	nt	frameshift with stop at codon 259
640 (2)	NSA	619^620ins9nt	LLR207-208ins	expansion of LRR6
R7 (7)	NSA	628^629ins9nt	CLR209-210ins	expansion of LRR6
P520 (2)	Netherlands 638T>A	638T>A	L213Q	missense, leucine to glutamine
580 (2)	. Canada	647A>G	N216S	missense, asparagine to serine
550 (9)(ref 28, family 3)	Germany	695T>C	L232P	missense, leucine to proline
B1 (3)	NSA	792C>G	N264K	missense, asparadine to Ivsine
B660 (1)	NSA	854T>C	L285P	missense, leucine to proline
B2 (5)	USA	893T>C	F298S	missense, phenylalanine to serine
610, 620 (4,10)	Costa Rica	1049G>A	W350X	protein truncation, loss of GPI-anchoring

a families Y1, Y2, Y3, R5, R7, B1 and B2 were first reported in reference 25. Earlier reports on other families is indicated following the family designation.

FIGURE 6

^bfollowing the recommendations of Nomenclature Working Group.

^cnumber of affected males in these families: 8, 1, 4, 8, 2 and 5, respectively. For all other families in this table this information is shown in first set of parentheses following the family designation.

din the analysis of X chromosomes with this deletion, we observed identical haplotypes across Xp11.4 from DXS556 to DXS228 with some chromosomes diverging proximally or distally beyond this region (15 markers tested) ^eLRR, leucine-rich repeat.

Patient 1, whose electrophysiological results are shown in Fig. 1.

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Ggctgagggagtggagggggacctcagaggagcaggaccaggggagactcccaggacggtaggggtcccacggctgggtggtcctaagccactgggtgg

- တ ပ ø O Ø O Ö 3 Ø Д ტ ы G × 1/1
- 121/41
- gcetteggeaegetgeegteettgegeegeetgtegetgegeeacaaacetgteetteateacgeeeggegeetteaagggeetgeeggeetgegetgagetgegeetagegeeacaac A ${ t F}$ G ${ t T}$ L ${ t P}$ S L R R L S L R H N N L S F I T P G A F K G ${ t I}$ P R ${ t I}$ A F ${ t I}$ R ${ t I}$ A ${ t R}$ ${ t R}$ ${ t R}$ ${ t I}$ A ${ t R}$ ${ t I}$ A ${ t R}$ ${ t R}$ ${ t I}$ A ${ t R}$ ${ t$ 241/81
- ggcgacctgcgctacctgcacgcgcgcaccttcgcgggcgctcagccgcctggcgcctagacctagcagcctcttcagcgtgcccgagcgctcctggccgaactgccggcc G D L R Y L H A R T F A A L S R L R R L D L A A C R L F S V P E R L L A E L P A 361/121
- 481/161
- ᄺ 601/201
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- gagctgctctacctggaccgcaacagcatcgccttcgtggaggaggcgcttccagaacctctcgggtctcctcgcgctgcacctcaacggcaaccgcctcaccgtgctcc $oldsymbol{ ext{E}}$ $oldsymbol{ ext{L}}$ $oldsymbol{ ext{L}}$ 841/281
- ۵ ۲ > ø G တ G ഥ Σ 3 Ω × Ц 3 Œ J α, ပ Ω ပ ပ 3 Д z ĸ Ŀ H Ĺ 1 α, G ы ĹŁ Ŀ G а ø Ĺ 961/321
- Δ, S ഗ H ⊱ H z Н (±) ш ىم Ω > ပ ᆸ G Ω ഗ ഗ ~ G [t_i ۲ > ø တ ᆸ Ω Ц G A > S G 4 S Æ O 1081/361
- 1201/401
- 1321/441
- 1441/481
- gtgggccctcggggtgggaagactaggaaagcttctaagggcttcacatcccttcccctcccctccccttccccttcatcttccaggcaacagtgcctgcaaggcctgaattagaaga **Gtttgcctccagagatggccccagggagacacagggacgtgccactcgagggggaggatggtatggtattgattctgtttgtcacacgggcatccattggaaaagagaagaagaagaatgaac** 1561 1681
 - ${\tt acttccalt}$ gotaagtagtiaagagccgtcccatttctcctggcggggtaacccaltacaccgaagtcctttgttttctaccacaatcctcctcctcctcctcgggggcctggaaacac 1801 1921
- 2041
 - cgaggattaaaaaaaaaaaaaggttaaaaccggtttctctctactgctggacttttcagagtctttc